

## EXPERT COMMENTARY

## Genetic Testing Offers Promise for Consumers

The expanding market for direct-to-consumer genetic testing raises important issues that physicians, patients, and policy makers are just starting to address. I consider DTC marketing of genetic information a natural evolution of technology, and a development that may offer benefits for individuals and society.

Consumers already have access to other channels for directly obtaining medical information about themselves, without consulting a health care provider. This is always a “buyer beware” situation, but not one that warrants regulations aimed at preventing consumers from using this emerging technology. Instead, I feel that policy efforts should be targeted toward these goals:

► Ensure the technical accuracy (analytic validity) of results obtained from DTC tests—whether genetic tests or not.

► Ensure that companies offering such services do not make unrealistic claims (clinical validity).

► Educate consumers about the limits of applying such information in clinical practice, especially when clinical studies have not been conducted.

► Educate health care providers about the clinical utility of such information, specifying what is and is not currently known.



BY HOWARD P. LEVY, M.D., PH.D.

My colleagues and I conducted a study of online resources that Joseph McNerney, executive director of the National Coalition for Health Professional Education in Genetics (NCHPEG), summarized at a National Academy

of Sciences workshop on DTC genetic testing (see article below). We identified a clear need for a point-of-care genetic education resource that is freely available, accurate, authoritative, and concise

and that allows users to find answers to their questions in less than 2 minutes (Genet. Med. 2008;10:659-67). Such a resource does not yet exist.

Our team is in the process of developing a resource of this kind, under the auspices of NCHPEG and in close collaboration with Mr. McNerney. We tentatively plan to launch the GeneFacts Web site in the fall of 2010.

Most companies offering DTC genetic testing claim or imply that knowledge of genetic risks will prompt consumers to engage in healthier behaviors and/or take appropriate medical action to mitigate their risks. This is wonderful in theory, but it has not yet been documented in the real world.

Genome-wide association studies and whole genome analysis are so new that we don't yet know how best to calculate an overall disease risk that incorporates multiple genetic risk factors. Even less is known about how to mathematically combine genetic risk factors for disease

with other, better-known risk factors such as family history, diet, and environmental exposures.

Despite the limitations in our knowledge of how to use newly identified genetic risk data, I do not think that access to DTC testing services should be restricted. I like the idea that the commercial marketing of whole genome analysis forces science and medicine to advance as rapidly as possible, to allow us to understand how best to use the information.

The potential benefit of DTC genetic testing comes at the cost of possibly harming individuals who use the information incorrectly, and that danger underscores the importance of education both for consumers and for health care providers. ■

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## Physicians Not Ready for Direct-to-Consumer Genetic Testing

BY JEFF EVANS

WASHINGTON — Few physicians feel prepared to interpret findings from direct-to-consumer genetic tests and incorporate the results into clinical practice, according to speakers at a National Academy of Sciences workshop on DTC genetic testing.

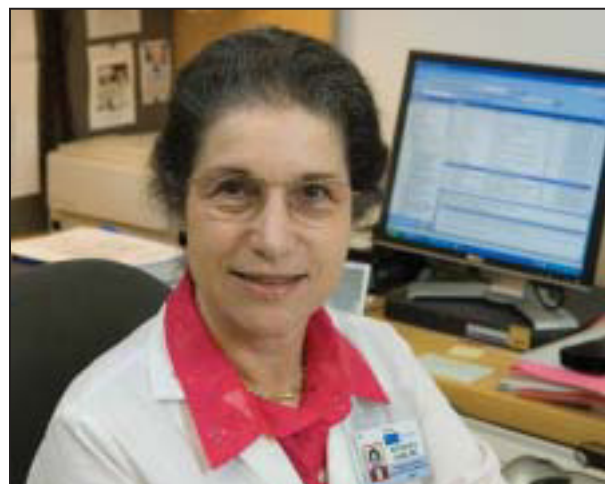
Surveys and anecdotal accounts discussed at the meeting cast doubt on the idea that physicians will be able to help consumers decide what to do about risks identified by DTC genetic tests.

“There's a lot of confusion between these services and medical care,” Dr. Patricia Ganz said. DTC companies may say that test results are for educational and research purposes only, and cannot be used for diagnostic purposes because the tests have not been validated for clinical use, but the results are “in fact being very

much treated as medical information.”

The difference between how the tests are marketed and what's feasible in clinical practice point to a “number of risks to the clinical encounter,” said Dr. Ganz, professor of health services and medicine at the University of California, Los Angeles. Possible problems include a demand for screening tests that have no proven clinical value, the perception that a physician is unsympathetic or lacking in knowledge when reviewing a patient's DTC genetic test report, and a false sense of security when a test result indicates “low risk.”

Some physicians currently use genetic tests with known clinical value, such as tests for blood disorders or prenatal risk assessment, Dr. Ganz said. But many physicians have little need for test results about cancer predisposition or other genetic syndromes, and are even less like-



“There's a lot of confusion between these services and medical care,” Dr. Patricia Ganz said.

ly to be prepared to interpret DTC genetic test reports that are derived from case-control association studies and genome-wide association studies.

Published reports indicate that physicians obtain most of their information about DTC genetic testing through the media, Katrina Goddard, Ph.D., of the Kaiser Permanente Center for Health Research, Portland, Ore., said at the workshop.

An online physician survey, called DocStyles, included 1,250 respondents (response rate 61%) in 2006 and 1,880 (response rate 22%) in 2008. More than 60% of the respondents reported getting information about DTC testing from the media, and less than 30% said they obtain information about such testing from other sources (Genet. Med. 2007;9:510-7; Genet. Med. 2009;11:595).

At the workshop, Joseph McNerney, executive director of the National Coalition for Health Professional Education in Genetics, said that individuals and fami-

lies with genetic conditions also do not appear confident about their provider's knowledge of genetics.

In a survey of 5,915 respondents conducted by the Genetic Alliance, an advocacy group, more than 30% rated as poor their provider's understanding of genetics and ability to deal with genetics-related management issues (Genet. Med. 2007;9:259-67).

Physicians who search for resources to help in interpreting

DTC test results are likely to turn to point-of-care clinical decision tools. But current versions of these tools often lack relevant information and are inefficient to use, Mr. McNerney said.

A study of two open-access and seven general-subscription genetics resources online found that in answering four clinical questions about each of five common genetic conditions, the resources provided complete information only one-third of the time, and in just as many instances provided no information (Genet. Med. 2008;10:659-67).

The investigators reported that their searches took 3-18 minutes to obtain a complete answer, which is longer than most physicians would be willing to spend, he said.

These results may reflect deficiencies in training in clinical genetics, he said. (See box.)

None of the speakers disclosed conflicts of interest with DTC genetic testing companies. ■

## Genetics Rare in Medical Schools

Only two U.S. medical schools have integrated medical genetics into their curricula for all 4 years, which suggests there are not enough professors and instructors sufficiently well trained in genetics to connect basic and clinical science during training, Mr. McNerney said at the workshop.

“There is a perception among many health care providers that genetics is still quite circumscribed by traditional, Mendelian, rare genetic disease and chromosomal anomalies, and that they are the province of primarily two different groups of providers—ob.gyns. and pediatricians,” he said.

He noted that a 2005 survey of 149 U.S. and Canadian course directors in medical genetics or curricular deans in medical schools found that 77% of the schools taught medical genetics in the first year, but only 47% incorporated it into the third or fourth year (Acad. Med. 2007;82:441-5).

The two schools with integrated genetics programs—the University of Vermont's Vermont Integrated Curriculum and Johns Hopkins University's Genes to Society program—both seek to teach how to evaluate human genetic variability in the context of the community and the environment.